Cystic Fibrosis

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Cystic fibrosis is quite a nasty genetic disease. It affects everyone differently. Its wide range of symptoms include a thick layer of mucus in the lungs which leads to much coughing and wheezing, often with blood streaking, an increased susceptibility to infections due to the layer of mucus which provides favorable breeding grounds for diseases, fatigue, digestive difficulties also due to excess mucus in the intestinal tracts. Other common characteristics are the loss of salt in sweat, asthma and abdominal pains. The average life expectancy for a person with cystic fibrosis is only 40-50 years, and this is in a situation with adequate healthcare.

One in 25 Caucasians are carriers of the defective CFTR gene, which causes cystic fibrosis, and around 30,000 Americans, 3,000 Canadians and 20,000 Europeans have cystic fibrosis. Cystic fibrosis also occurs in Asia and Africa, but it is most common in countries where the inhabitants are of Northern European ancestry.

Cystic fibrosis is caused by a mutation of the CFTR gene, which is located on the longer arm of the 7th chromosome. This gene controls the movement of salt and water in and out of cells, but when it is defective it has difficulties doing so, resulting in all the sticky mucus in the lungs, intestines and pancreas. The dysfunctional CFTR gene is an autosomal recessive trait, meaning that two of the parents must be either carriers or sufferers of the disease for the child to have the defective gene.

It is possible to detect the gene for cystic fibrosis before the birth of the baby by carrying out genetic tests on fluid from the mother’s womb or cells from the tissue that will form the placenta. However this can be risky for the pregnant mother, so it is only done in cases where there is another child in the family with cystic fibrosis.

Currently, there is no actual cure for cystic fibrosis, though gene replacement or transplantation may be an option for the near future. However, there are many methods to alleviate the symptoms. Exercise is always a good option as it helps cough up the mucus from the lungs. There are also a wide range of medicines to ease the breathing such as ‘mucolytics’ which diminish the mucus in the lungs, and decongestants which reduce the swelling or inflammation in the lungs. A more traditional method is for the sufferer to get into a specific position, and then be pounded on different places on the back to get the mucus out so the sufferer can cough it up. Often, diets are prescribed to the patients so that they will encounter less digestive difficulties.

 There are lots of upcoming medicines or techniques, which help alleviate

the symptoms of cystic fibrosis. One of the more recent examples of this would be a new drug called VX-770, pictured on the left. It addresses the impaired CFTR protein, and improves chloride transport, leading to improvement in lung function, and a reduction in salt lost in sweat.

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